

Sequence Variant Nomenclature

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HGVS nomenclature activities performed in 2022

Rotterdam January 2023
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Short report on the main activities in 2022 by the “HGVS Variant Nomenclature Committee” (HVNC), a committee working under the auspices of the Human Genome Organization (HUGO), as a member of the HUGO Nomenclature Standards Committee (HNSC). The activities concern the topic of HGVS nomenclature (the “HGVS recommendations for the description of sequence variants”). The report is written on behalf of the HVNC.

HGVS Variant Nomenclature Committee

(previous called the Sequence Variant Description working group (SVD-WG))

- early 2022 a call was published for new members of the HVNC. The HVNC evaluated all applications and selected four to be nominated by the HUGO board. The HUGO board approved the nominations and Alex Wagner (USA), Rachel Taylor (UK), Marina DiStefano (USA) and Jeroen Laros (NL) were elected as new HVNC member for a 4-year period from 2022-2025.
- members of the new committee met for the first time in an online meeting (Oct.2022). The main topic discussed was what the members considered the best way to work and communicate. Several options were put forward to be discussed further at a next meeting (January 2023).
- Raymond Dagleish (Leicester, UK), a member since the start of the nomenclature committee, decided to step down from the committee. The HVNC re-evaluated the applications received and, as proposed by Raymond Dagleish, suggested the HUGO board to nominate Ivo Fokkema (NL) as HVNC member. The HUGO board approved the nomination and Ivo Fokkema was elected as HVNC member for a 4-year period from 2023-2026.
- no new versions of the HGVS nomenclature were released. The current version of the HGVS nomenclature remains v20.05.

Promotion

- HGVS online Q&A

The bi-monthly online HGVS nomenclature Q&A sessions started in June 2021 were ended in January. Although participants following the sessions live appreciated the format very much, too few attended to make the efforts and cost involved worthwhile. Questions discussed are still available as Socrative

classrooms (www.Socrative.com), and users still use these to test their knowledge on HGVS nomenclature.

- VarNomen@HGVS.org e-mail

The chair received ~120 questions about HGVS nomenclature by e-mail, all of which have been answered.

- HGVS nomenclature Facebook

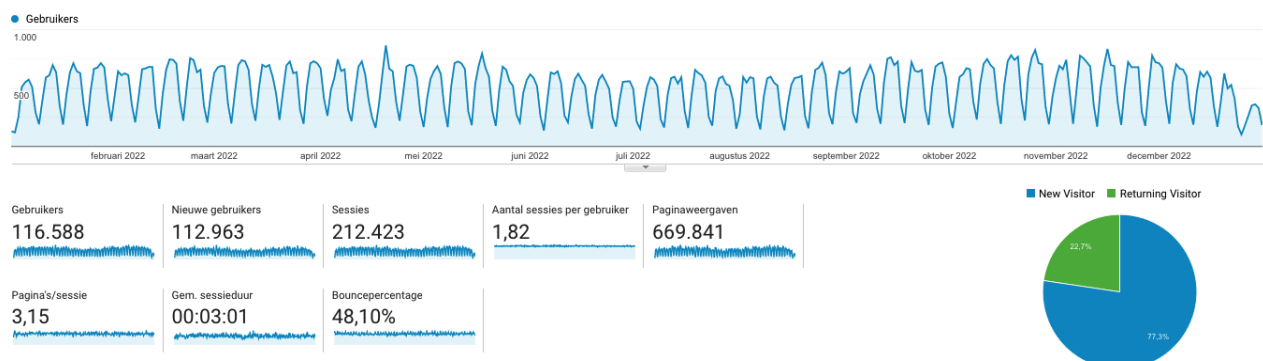
Based on the e-mail questions received, 15 posts (26 in 2020) were made on Facebook. In addition, some 10 questions received on Facebook were answered.

- VarNomen.HGVS.org website

Many improvements were made to the HGVS nomenclature website (mainly correcting mistakes, adding examples, general updates), most triggered by questions or remarks received by e-mail.

- web site statistics

The HGVS nomenclature website visits stabilised when compared to 2021. Daily visits peaked at 867 in April (2020 peak 783). Annual totals for 2022 are 669,841 pageviews (692k in 2021) in 212,423 sessions (209k in 2021), from 116,588 visitors (118k in 2021).



Education

- a workshop on HGVS nomenclature was presented during the GV/HUGO “Variant Effect Prediction training course”.

- the European Society of Human Genetics (ESHG) decided in 2019 to start with the organisation of so-called pre-meeting courses. Because of the COVID pandemic, the courses could so far no be organised. The first pre-meeting courses will now start on Friday June 9, 2023. One of the pre-meeting courses offered will be “HGVS nomenclature”, from 12.00-17.00, organised by Johan den Dunnen.

Current members HGVS Variant Nomenclature Committee (HVNC)

- Johan den Dunnen (chair)
- Ros Hastings (ISCN)
- Peter Causey-Freeman
- Reece Hart
- Terence Murphy
- Jeroen Laros
- Ivo Fokkema
- Rachel Taylor
- Marina DiStefano
- Alex Wagner